AAAAI Ask the Expert
Possible diagnosis of Type III hereditary angioedema

2/22/2012

Q: I have an 11 yo female patient with a 2 month history of (mild) lip and throat swelling but more recently impressive abdominal distension/pain for the past 5 days. She did not respond to H1/H2 or po steroids and C1 esterase levels and fx were nl during an attack (C4 low but not yet repeated and C2 still pending). Abd CT and GI neg except for some mild/mod stool that was cleaned out and she still has sx. I am considering Type III angioedema since there is no other explanation but what would be the best (most effective and safest) tx option to try and get her insurance to cover given her age? Thank you.

A: The index of suspicion of Type III hereditary angioedema is sharpened by a positive family history. In the absence of a family history, I would not suspect this condition, and a diagnosis cannot be established. In addition, a CT scan of the abdomen taken during an episode of angioedema would be very likely to show clearcut findings with gastrointestinal angioedema being quite evident. If the CT scan was taken during the episode, and no angioedema was noted, I would pursue another cause for her abdominal symptoms. The presence of abdominal symptoms for five days would also be a little unusual for angioedema as the cause.

As you know, the vast majority of episodes of angioedema of the lip and throat are idiopathic, and if she has had clearcut visual evidence for angioedema of the lip with the remainder of the clinical picture you describe, I would think that idiopathic angioedema would be the most likely diagnosis in your patient. In the absence of any objective findings of a complement abnormality and abdominal angioedema on CT scan, I would not think that treatment for hereditary angioedema such as the administration of C1 esterase inhibitor, Ecallantide, Icatibant, or modified steroids would be at this time indicated.

However, I am going to ask Dr. Bruce Zuraw, who, as you know, is an international expert in angioedema involving the complement system, to share his thoughts with us. When I hear from Dr. Zuraw, I will forward his response to you.

Thank you again for your inquiry.

Sincerely,
Phil Lieberman, M.D.

We received a response from Dr. Bruce Zuraw. Thank you again for your inquiry and we hope this response is helpful to you.

Sincerely,
Phil Lieberman, M.D.

Response from Dr. Bruce Zuraw:
I agree with your response. One cannot make a diagnosis of type III in this situation. Without a test to rule-in type III HAE it is necessary to have a clear personal and family history of recurrent angioedema, recognizing that there is the possibility of missing the diagnosis in some patients.

Similarly, I would agree that the CT ought to show the edema if that was the problem. I do note that the C4 is reported as low, and this is a more accurate assay for HAE than either C1 inhibitor antigen or C1 inhibitor function by the EIA assay (most commonly available in the USA with normal reported as >67%). I'd repeat the C4 and also obtain a repeat C1 inhibitor functional assay using the chromogenic assay (available from National Jewish Labs) in case this is a type II HAE patient.

Regards,
Bruce Zuraw